

## NPTR VIDEO CATALOG ENTRY

Disease	Homocystinuria (HCU). Amino acid disorders.
Title	Homocystinuria Patient Testimony - Joseph.
Link	<a href="https://www.youtube.com/watch?list=PLvz3MWdu8TP3wTE9wYNxq3WOxQxMbx4XV&amp;v=HpCzhWS7iWI&amp;feature=emb_title">https://www.youtube.com/watch?list=PLvz3MWdu8TP3wTE9wYNxq3WOxQxMbx4XV&amp;v=HpCzhWS7iWI&amp;feature=emb_title</a>
Key words	Homocystinuria (HCU). Amino acid disorders. Patient/family experience. Symptoms. Diet/treatment.
Description	Joseph was diagnosed late with homocystinuria. He describes the challenges in living with this disorder, including the treatment and its demanding rigors, insurance coverage, etc. Aspects of Joseph's neurologic phenotype can be viewed (note his expressive speech pattern and speech content), as well as his aphakic lenses.
Length (min:sec)	6.14
Speaker Background	Patient.
Objectives	<ol style="list-style-type: none"> <li>1. Discuss some of the day-to-day challenges/experiences of a patient living with homocystinuria.</li> <li>2. Describe the clinical impact of homocystinuria.</li> </ol>
Educational utility	Provides a personal narrative about living with the disease but should be accompanied by significant medical teaching to increase understanding about the disorder. Notable aspects of his presentation can be highlighted to viewers so they better appreciate the phenotype.
Technical features	Great audio and video quality.
Relevant/ Target Audience	Health professionals and specialists. Trainees and students in the health professions. Other stakeholders who have an interest in the inborn errors.
Promotional aspects	The patient mentions his formula product and medication. This video does not otherwise promote a commercial product, treatment, and/or medical device.
Source	HCU Network America.

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